



## PKD2 gene

polycystin 2, transient receptor potential cation channel

### Normal Function

The *PKD2* gene provides instructions for making a protein called polycystin-2. This protein is found in the kidneys before birth and in many adult tissues. Although its exact function is not well understood, polycystin-2 can be regulated by a larger, somewhat similar protein called polycystin-1.

Polycystin-2 likely functions as a channel spanning the cell membrane of kidney cells. In conjunction with polycystin-1, the channel transports positively charged atoms (ions), particularly calcium ions, into the cell. This influx of calcium ions triggers a cascade of chemical reactions inside the cell that may instruct the cell to undergo certain changes, such as maturing to take on specialized functions. Polycystin-1 and polycystin-2 likely work together to help regulate cell growth and division (proliferation), cell movement (migration), and interactions with other cells.

Polycystin-2 is also active in other parts of the cell, including cellular structures called primary cilia. Primary cilia are tiny, fingerlike projections that line the small tubes where urine is formed (renal tubules). Researchers believe that primary cilia sense the movement of fluid through these tubules, which appears to help maintain the tubules' size and structure. The interaction of polycystin-1 and polycystin-2 in renal tubules promotes the normal development and function of the kidneys.

### Health Conditions Related to Genetic Changes

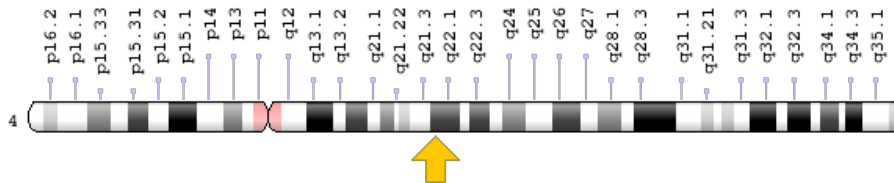
#### polycystic kidney disease

More than 75 mutations in the *PKD2* gene have been identified in people with polycystic kidney disease. These mutations are responsible for about 15 percent of all cases of autosomal dominant polycystic kidney disease (ADPKD), which is the most common type of this disorder. Mutations in the *PKD2* gene include changes in single DNA building blocks (base pairs) and deletions or insertions of a small number of base pairs in the gene. Most *PKD2* mutations are predicted to result in the production of an abnormally small, nonfunctional version of the polycystin-2 protein. Although researchers are uncertain how a lack of polycystin-2 leads to the formation of cysts, it likely disrupts the protein's interaction with polycystin-1 and alters signaling within the cell and in primary cilia. As a result, cells lining the renal tubules may grow and divide abnormally, leading to the growth of numerous cysts characteristic of polycystic kidney disease.

## Chromosomal Location

Cytogenetic Location: 4q22.1, which is the long (q) arm of chromosome 4 at position 22.1

Molecular Location: base pairs 88,007,647 to 88,077,779 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- APKD2
- Pc-2
- PC2
- PKD2\_HUMAN
- PKD4
- polycystic kidney disease 2 (autosomal dominant)
- polycystin-2
- TRPP2

## Additional Information & Resources

### GeneReviews

- Polycystic Kidney Disease, Autosomal Dominant  
<https://www.ncbi.nlm.nih.gov/books/NBK1246>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PKD2%5BTIAB%5D%29+OR+%28polycystic+kidney+disease+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

## OMIM

- POLYCYSTIN 2  
<http://omim.org/entry/173910>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_PKD2.html](http://atlasgeneticsoncology.org/Genes/GC_PKD2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=PKD2%5Bgene%5D>
- HGNC Gene Family: EF-hand domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/863>
- HGNC Gene Family: Transient receptor potential cation channels  
<http://www.genenames.org/cgi-bin/genefamilies/set/249>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=9009](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9009)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/5311>
- UniProt  
<http://www.uniprot.org/uniprot/Q13563>

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